



RETREG1 gene

reticulophagy regulator 1

Normal Function

The *RETREG1* gene provides instructions for making a protein important for the survival of sensory and autonomic nerve cells (neurons). Sensory neurons transmit pain, touch, and temperature sensations. Autonomic neurons help control involuntary functions of the body such as heart rate and blood pressure. Within neurons, the RETREG1 protein is found in a structure called the Golgi apparatus, which is important for distribution of proteins to the cell. However, the function of the RETREG1 protein is not well understood. Studies have shown that neurons in which the RETREG1 protein is absent die by a process of self-destruction called apoptosis.

Health Conditions Related to Genetic Changes

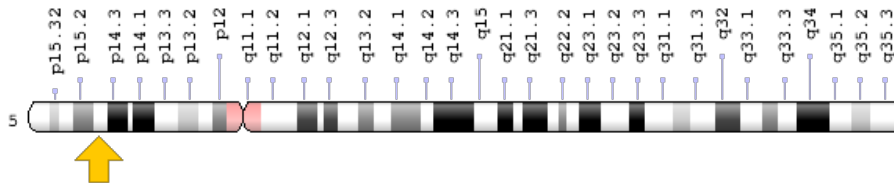
hereditary sensory and autonomic neuropathy type II

Mutations in the *RETREG1* gene are responsible for one type of hereditary sensory and autonomic neuropathy type II (HSAN2) called HSAN2B. People with HSAN2B lose the ability to feel pain or sense hot and cold. The *RETREG1* gene mutations may lead to an abnormally short and nonfunctional protein. The lack of RETREG1 protein causes neurons to undergo apoptosis, which reduces the overall number of sensory and autonomic neurons. The loss of neurons results in the signs and symptoms of HSAN2B.

Chromosomal Location

Cytogenetic Location: 5p15.1, which is the short (p) arm of chromosome 5 at position 15.1

Molecular Location: base pairs 16,473,038 to 16,617,058 on chromosome 5 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- F134B_HUMAN
- FAM134B
- FAM134B protein
- FAM134B protein isoform 1
- FAM134B protein isoform 2
- family with sequence similarity 134 member B
- family with sequence similarity 134, member B
- FLJ20152
- FLJ22155
- FLJ22179
- JK1

Additional Information & Resources

GeneReviews

- Hereditary Sensory and Autonomic Neuropathy Type II
<https://www.ncbi.nlm.nih.gov/books/NBK49247>

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28FAM134B%5BTIAB%5D%29+OR+%28JK1%5BTIAB%5D%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D>

OMIM

- FAMILY WITH SEQUENCE SIMILARITY 134, MEMBER B
<http://omim.org/entry/613114>

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
http://atlasgeneticsoncology.org/Genes/GC_FAM134B.html
- ClinVar
<https://www.ncbi.nlm.nih.gov/clinvar?term=RETREG1%5Bgene%5D>
- HGNC Gene Symbol Report
http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=25964
- NCBI Gene
<https://www.ncbi.nlm.nih.gov/gene/54463>
- UniProt
<http://www.uniprot.org/uniprot/Q9H6L5>

Sources for This Summary

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Reprinted from Genetics Home Reference:
<https://ghr.nlm.nih.gov/gene/RETREG1>

Reviewed: May 2011
Published: April 25, 2017

Lister Hill National Center for Biomedical Communications
U.S. National Library of Medicine
National Institutes of Health
Department of Health & Human Services